LABORATORY BULLETIN

DEPARTMENT OF HEALTH & ENVIRONMENTAL SCIENCES, HELENA, MONTANA

No. 53 David B. Lackman, Ph.D., Administrator, Laboratory Division April 1, 1974

AMENDMENT OF RULE MAC 16-2.18 (6)-S1820 (PKU TESTS, INFANTS)

Sections 69-6710 through 69-6713, R.C.M. (House Bill 261, 1973 session) authorize the Department to amend administrative codes governing PKU testing of infants to include screening tests for other inborn errors of metabolism. Pursuant to the provisions of this act, an advisory committee was convened in Helena on January 11, 1974, to make recommendations for amendment of this rule. The consensus from this meeting formed the basis for proposed amendments published in advance of a public hearing held on March 19, 1974. At this hearing before the Board of Health and Environmental Sciences, testimony was presented which resulted in additional changes. The Rules as adopted by the Board of Health and Environmental Sciences on March 19, 1974 follow:

MONTANA ADMINISTRATIVE CODE pp 16-404 to 16-406, Sub-Chapter 6,
Maternal and Child Health Bureau: 16-2.18 (6)-S1820 INFANT SCREENING TESTS

(1) Persons in charge of any facility caring for newborn infants and persons responsible for the registration of births shall ensure that each infant has tests for inborn errors of metabolism. These shall include a test for phenylketonuria and tests for detection of other aminoacidopathies.

(2) Only laboratory tests approved by the department shall comply with the provisions of sections 69-6710 through 69-6713, R.C.M. 1947.

(3) A laboratory must be approved by the department in order to comply with the provisions of this rule. Such laboratory shall report all positive or suspicious test results to the department within 48 hours after drawing the blood and performing the test.

(4) A newborn is an infant 28 days old or less.

(5) Required specimens for testing shall be taken by the hospital or institution wherein newborn care was rendered on the third day of life or 48 hours following ingestion of milk but not later than the 14th day of life.

(a) In the event the newborn is discharged from the hospital prior to the third day of life, the tests shall not be performed before discharge. In this case, it shall be the duty of the administrative officer or other person in charge of each hospital or institution caring for newborn infants to make provision at the time of discharge for the proper testing of the newborn and to explain the reasons why it is of utmost importance to return for these tests. The parent or legal guardian of the newborn shall also be required to sign a statement assuming responsibility to cause the tests to be administered between the third and 14th day of life.

(6) Premature infants:

(a) A sample of blood at one to two weeks or discharge, whichever is earlier, shall be taken. If under 5-1/2 pounds at two weeks and initial sample is negative, repeat once more at discharge.

(b) Prematures with phenylalanine levels of 20 mg. percent or higher if found to have a high blood level of phenylalanine without elevation of the serum tyrosine levels may be given a provisional low protein diet which restricts phenylalanine intake somewhat but meets the requirements of the infant. These patients shall be challenged with phenylalanine intake while carefully monitoring their phenylalanine levels when they are about 5-1/2 pounds in order to establish a discarded diagnosis

(7) In the event of transfer of newborn infant to another hospital or institution, the tests shall be performed by the transferring hospital if transfer occurs on or after the third day of life and by the receiving hospital or institution if the transfer occurs before the third day of life.

(over)

The hospital or institution receiving a newborn which was not previously tested shall perform the required tests between the third and 14th day of life unless medically contraindicated, in which case the tests shall be performed as soon as the medical condition of the infant permits such testing.

(8) When an infant has been born outside of a hospital or institution and has not subsequently been admitted to an institution for initial newborn care, it shall be the duty of the person required in section 69-4413, R.C.M. 1947, to register the birth of a child to assume responsibility to cause the tests to be administered not later than the 14th day of life, unless medically contraindicated, in which case they shall be performed as soon as the medical condition of the infant permits.

(9) An infant who has a positive or suspicious initial test shall immediately have a second test performed. If the second test is positive or suspicious, a blood specimen will be sent to a laboratory qualified to perform quantitative analysis for the substance in question.

(10) The administrator of the responsible hospital or institution and the person required to register the birth of a child shall:

(a) Be certain, prior to the discharge of an infant, that the specimens to be forwarded to the laboratory are adequate for testing purposes.

(b) Within 24 hours after the taking of the specimen, cause such specimen to be forwarded to the designated laboratory by first class mail or its equivalent.

(c) Cause to be recorded on the infant's chart the date of taking of the test specimen.

(d) Cause the result of the tests, as prescribed by the department, to be recorded on the infant's hospital chart and reported to the attending physician.

The sense of the meeting of January 11, 1974 was that:

a. Screening of newborn for evidence of phenylketonuria should be continued as currently carried out.
b. Screening for other aminoacidopathies should be added.
c. Physicians should be encouraged to have a urine sample tested in the hospital for mucopolysaccharides and for evidence of galactosemia.
d. In cases of prolonged jaundice (over four days), determination of the free thyroxine index should be done for detection of congenital hypothyroidism.

There are two qualifications concerning these recommendations which deserve mention:

1. Some have recommended the routine screening of newborn for evidence of inherited hypothyroidism by doing T_4 and T_3 tests. I requested an opinion on this from consultants at the Center for Disease Control and here is a statement received from Samuel Goldberg, M.D., in a letter dated November 6, 1972.

"It is our belief that the use of T_3 and T_4 screening as a means of preventing the development of cretinism would not be useful in newborns. It is difficult to make the diagnosis of cretinism before the age of 2 months and chemical parameters do not become apparent due to transplacental transport of one hormone from the mother at least until several weeks after birth in most cases. Certainly the screening of all infants with prolonged physiological jaundice might be deemed useful as these may yield a significant number of cases." *

2. In reviewing information from other states, I note that the test most frequently aded when PKU programs are expanded to include other genetic errors in metabolism is a screening test for galactosemia. This is usually done on another disc from the filter paper by the Beutler fluorometric method. However, the advisory committee considered it desirable to have a test for galactose done on a urine sample taken in the

hospital; and the same for mucopolysaccharides. There are dipsticks available for these tests but they require more evaluation. The Barry spot test with ortho-toluidine reagent is also used for mucopolysaccharides.

Presently we are working out the logistics of collecting specimens required to comply with these amendments. In addition to the blood spots on filter paper for the Guthrie PKU test, two capillary tubes of blood will be needed. Although some laboratories in Montana already offer the tests added by the amendments, most of the testing will probably be done under contract with a consulting laboratory.

REPORTING

One of the greatest operating handicaps plaguing our Preventive Health Services Bureau is failures in reporting cases of disease. Often the first and only indication received of the occurrence of a reportable disease is a duplicate copy of reports of positive findings in the laboratory. It should not be this way because our reports often are delayed. Repetition of a comment from Dr. Skinner in Communicable Disease Notes for December 5, 1973, is in order. *"The responsibility for communicable disease reporting and control rests with all of us, not only with the medical community. The regulations of the Montana State Board of Health state that any person having knowledge of a reportable, communicable disease should report such knowledge to the local health officer. If the person reporting is not a physician, the local health officer must verify the diagnosis. If anything is to be accomplished in protecting Montana people against epidemic diseases, we must know that the disease is present."*

REMINDER

While we are dealing with rules and regulations, here is a reminder from MONTANA LICENSING LAW AND REGULATIONS FOR HOSPITALS AND RELATED INSTITUTIONS, January 1, 1972, Section 31.105 Laboratory pg. 17

"e. The laboratory should be under the full-time direction of a pathologist or should have the services of such a person as a consultant. A consultant should make monthly visits as a minimum. If the laboratory is not under the full-time direction of a pathologist, a staff physician or qualified laboratory technologist (preferably registered with a nationally recognized accrediting body) or a laboratory specialist qualified by a doctoral degree should be named to be responsible for the proper operation of the laboratory."

* Another important consideration when looking for inborn errors of metabolism is family history; especially one of thyroid disease in the mother.

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